HLA CELIAC DISEASE
(DQB1*02,1*03,DQA1*05;DQA1:03)
WHOLE BLOOD
(PCR – SSP)

Positive /
Negative

DRB1*03 – DQA1*05:01 –
DQB1*02:01{DQ 2 (DQ 2.5)}

DRB1*07 – DQA1*02:01 –
DQB1*02:02 (DQ 2)

DRB1*11 – DQA1*05:05 –
DQB1*03:01(DQ 7)

DRB1*04 – DQA1*03:01 –
DQB1*03:02(DQ 8)

Note: Results to be correlated with Serological studies / Biopsy

Comment

DQ 2.5 (DRB1*03 – DQA1*05:01 – DQB1*02:01) represents the highest risk for Celiac Disease which is five times higher if it is homozygous. The associated risk is also high if DQ2.5 – DQ 8 combination is present, but is lower with DQ2 (DRB1*03 – DQA1*05:01 – DQB1*02:02) and needs to be correlated clinically. DQ 8 alone is found in 2-10 % of patients with Celiac Disease.

The diagnosis of Celiac Disease (CD) is based on a combination of history and clinical presentation, serological tests (Tissue –Transglutaminase or Anti Endomysial antibody) and small intestine biopsy. Screening for HLA –DQ 2 and DQ 8 has low specificity and positive predictive value as approximately 30% and 20% respectively of healthy population may test positive for these alleles. The test has excellent negative predictive value and can be assumed that in more than 90% cases Celiac Disease does not exist. The incidence of CD is 10-20 fold that of general population in first degree relatives of a patient. It is also 16-20 times higher in cases of Type 1 Diabetes mellitus and Down's Syndrome. These cases may be screened by this assay rather than serological testing at regular intervals.